Open Peer Review on Qeios

Ornithine Carbamoyltransferase Deficiency Disease

National Cancer Institute

Source

National Cancer Institute. <u>Ornithine Carbamoyltransferase Deficiency Disease</u>. NCI Thesaurus. Code C84957.

An X-linked urea cycle metabolic disorder characterized by deficiency of ornithine carbamoyltransferase, resulting in the accumulation of amino acids and ammonia in the serum. Signs and symptoms include seizures, delayed growth, behavioral changes, ataxia, lethargy, and coma.